The proband, a 23-year-old Brazilian female patient, is the third child of healthy, non-consanguineous parents. Her birth weight was 1,900 g, the length was 47 cm, the occipitofrontal circumference (OFC) was 36 cm. Developmental milestones were delayed: she sat without support at the age of 1, walked at 2.5 years and acquired control of the bowels at age 4. She spoke only a few meaningful words at the age of eighteen. The feeding problems were noted in infancy.

A physical examination at 23 years of age showed a weight of 61 kg (75<p<97.5), a height of 139.5 cm (p<2.5) and an OFC of 49.5 cm (p<2.5). Pertinent physical characteristics were highly arched eyebrows, downward-slanted palpebral fissures, a beaked nose, nasal septum below alae, a highly arched palate and posteriorly angled ears. She also had broad, radially deviated thumbs, broad great toes and long halluces.